SKIN DISORDERS GENE PANEL DG 2.4.x

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
AAAS	92	100%	100%	Achalasia-addisonianism-alacrimia syndrome,231550
AAGAB	109.9	100%	95%	Keratoderma palmoplantar punctate type IA,148600
ABCA12	100.8	100%	99%	Ichthyosis, autosomal recessive 4B (harlequin),242500 Ichthyosis, congenital, autosomal recessive 4A,601277
ABCB6	115.8	100%	100%	Dyschromatosis universalis hereditaria 3,615402 Microphthalmia,isolated, with coloboma 7,614497 [Blood group, Langereis system],111600
ABCC6	50.8	70%	67%	Arterial calcification generalized of infancy 2,614473 Pseudoxanthoma elasticum,264800 Pseudoxanthoma elasticum, forme fruste,177850
ABCC9	107	99%	97%	Atrial fibrillation familial 12,614050 Cardiomyopathy, dilated, 1O,608569 Hypertrichotic osteochondrodysplasia, 239850
ABHD5	116.3	100%	95%	Chanarin-Dorfman syndrome,275630
ACTA2	82.9	100%	99%	Aortic aneurysm familial thoracic 6,611788 Moyamoya disease 5,614042 Multisystemic smooth muscle dysfunction syndrome,613834
ACVRL1	53.8	94%	88%	Telangiectasia hereditary hemorrhagic type 2,600376
ADAM10	118.1	100%	100%	Reticulate acropigmentation of Kitamura,615537 {Alzheimer disease 18, susceptibility to},615590
ADAM17	111.6	99%	97%	Inflammatory skin and bowel disease neonatal,614328
ADAMTS10	73.4	97%	93%	Weill-Marchesani syndrome 1 recessive,277600
ADAMTS17	73.9	92%	81%	Weill-Marchesani-like syndrome,613195
ADAMTS2	98.3	98%	94%	Ehlers-Danlos syndrome type VIIC,225410
ADAR	128.1	99%	98%	Aicardi-Goutieres syndrome 6,615010 Dyschromatosis symmetrica hereditaria,127400
AGA	111.6	100%	91%	Aspartylglucosaminuria,208400

AGPAT2	60.9	90%	85%	Lipodystrophy congenital generalized type 1,608594
AIRE	73.4	99%	89%	Autoimmune polyendocrinopathy syndrome type I with or without reversible metaphyseal dysplasia,240300
AKT1	108.4	97%	95%	Breast cancer somatic,114480 Colorectal cancer, somatic,114500 Cowden syndrome 6,615109 Ovarian cancer, somatic,167000 Proteus syndrome, somatic,176920 {Schizophrenia, susceptibility to},181500
AKT3	103.7	100%	100%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome,603387
ALAD	91.9	96%	90%	Porphyria acute hepatic,612740 {Lead poisoning, susceptibility to},612740
ALAS2	84.4	92%	88%	Anemia sideroblastic X-linked,300751 Protoporphyria, erythropoietic, X-linked,300752
ALDH18A1	97.7	99%	95%	Cutis laxa autosomal recessive type IIIA,219150
ALDH3A2	93.7	100%	100%	Sjogren-Larsson syndrome,270200
ALDOB	117.9	100%	99%	Fructose intolerance,229600
ALOX12B	96	100%	99%	Ichthyosis congenital autosomal recessive 2,242100
ALOXE3	85.3	100%	98%	Ichthyosis congenital autosomal recessive 3,606545
ALPL	80.9	100%	96%	Hypophosphatasia, adult,146300 Hypophosphatasia, childhood,241510 Hypophosphatasia, infantile, 241500 Odontohypophosphatasia,146300
ALX4	72	100%	99%	Frontonasal dysplasia 2,613451 Parietal foramina 2,609597 {Craniosynostosis 5, susceptibility to},615529
AMELX	119.1	100%	100%	Amelogenesis imperfecta, type 1E,301200
ANKRD11	105.4	91%	86%	KBG syndrome,148050
ANTXR1	75.6	96%	90%	GAPO syndrome,230740 {Hemangioma, capillary infantile, susceptibility to},602089
ANTXR2	110.1	100%	97%	Hyaline fibromatosis syndrome,228600
AP3B1	104.8	100%	99%	Hermansky-Pudlak syndrome 2,608233

APC	144.1	100%	99%	Adenoma,periampullary,somatic,175100 Adenomatous polyposis coli,175100 Brain tumor-polyposis syndrome 2,175100 Colorectal cancer,somatic,114500 Desmoid disease,hereditary,135290 Gardner syndrome,175100 Gastric cancer,somatic,613659 Hepatoblastoma,s
APCDD1	119.5	100%	100%	Hypotrichosis 1,605389
AQP5	94.1	100%	94%	Palmoplantar keratoderma, Bothnian type,600231
ARHGAP31	135.6	100%	98%	Adams-Oliver syndrome 1,100300
ARID1A	102.4	99%	96%	Mental retardation autosomal dominant 14,614607
ARID1B	107	99%	96%	Mental retardation,autosomal dominant 12,614562
ASIP	63.5	72%	59%	[Skin/hair/eye pigmentation 9],611742
ASL	72.8	97%	93%	Argininosuccinic aciduria,207900
ASXL1	141.6	98%	97%	Bohring-Opitz syndrome,605039 Myelodysplastic syndrome,somatic,614286
ASXL3	148.6	100%	99%	Bainbridge-Ropers syndrome ,615485
ATIC	109.4	100%	97%	AICA-ribosiduria due to ATIC deficiency,608688
ATP2A2	115.4	100%	100%	Acrokeratosis verruciformis,101900 Darier disease,124200
ATP2C1	113.4	100%	99%	Hailey-Hailey disease,169600
ATP6V0A2	97.8	100%	99%	Cutis laxa,autosomal recessive,type IIA,219200 Wrinkly skin syndrome,278250
ATP7A	122.6	100%	100%	Menkes disease,309400 Occipital horn syndrome,304150 Spinal muscular atrophy,distal,X-linked,300489
ATR	113.1	100%	99%	Cutaneous telangiectasia and cancer syndrome familial,614564 Seckel syndrome 1,210600
AXIN2	95.6	97%	91%	Colorectal cancer somatic,114500 Oligodontia-colorectal cancer syndrome,608615
B3GALT6	56.2	79%	73%	Ehlers-Danlos syndrome progeroid type 2,615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1,with or without fractures,271640
B4GALT7	77.9	100%	96%	Ehlers-Danlos syndrome progeroid type 1,130070

BANF1	44.6	55%	54%	Nestor-Guillermo progeria syndrome,614008
BAP1	89.6	99%	97%	Tumor predisposition syndrome,614327
BCOR	124.7	100%	99%	Microphthalmia syndromic 2,300166
BCS1L	144	100%	100%	Bjornstad syndrome,262000 GRACILE syndrome,603358 Leigh syndrome,256000 Mitochondrial complex III defiency, nuclear type 1,124000
BLM	114.8	100%	98%	Bloom syndrome,210900
BLOC1S3	33.9	83%	73%	Hermansky-Pudlak syndrome 8,614077
BLOC1S6	118.7	88%	81%	Hermansky-pudlak syndrome 9,614171
BMS1	37.9	37%	36%	Aplasia cutis congenita, nonsyndromic,107600
BRAF	72.6	100%	97%	Adenocarcinoma of lung,somatic,211980 Cardiofaciocutaneous syndrome,115150 LEOPARD syndrome 3,613707 Noonan syndrome 7,613706
BRIP1	120.9	100%	99%	Breast cancer early-onset,114480 Fanconi anemia,complementation group J,609054
BSCL2	103.8	100%	100%	Encephalopathy, progressive, with or without lipodystrophy,615924 Lipodystrophy congenital generalized type 2,269700 Neuropathy, distal hereditary motor, type VA,600794 Silver spastic paraplegia syndrome,270685
BTD	130.2	100%	100%	Biotinidase deficiency,253260
C10orf11	89.8	99%	99%	Albinism, oculocutaneous type VII,615179
C1QA	119.1	98%	91%	C1q deficiency,613652
C1QB	90.3	94%	87%	C1q deficiency,613652
C1QC	121.9	84%	69%	C1q deficiency,613652
C2CD3	107.5	95%	95%	?Orofaciodigital syndrome XIV,615948
C4orf26	126.2	100%	100%	Amelogenesis imperfecta, type IIA4,614832
CA2	146.1	100%	100%	Osteopetrosis,autosomal recessive 3,with renal tubular acidosis,259730
CARD14	63	97%	87%	Pityriasis rubra pilaris,173200 Psoriasis 2,602723
CARD9	64.3	99%	98%	Candidiasis,familial 2,autosomal recessive,212050
CAST	95.4	98%	97%	PLACK syndrome,616295

CAV1	139.5	100%	100%	?Lipodystrophy,congenital generalized,type 3,612526 ?Partial lipodystrophy, congenital cataracts and neurodegeneration syndrome,606721 Pulmonary hypertension, primary, 3,615343
CBL	121.9	100%	100%	Noonan syndrome-like disorder,with or without juvenile meylomonocytic leukemia,613563
CBS	76.4	99%	81%	Homocystinuria B6-responsive and nonresponsive types,236200 Thrombosis,hyperhomocysteinemic,236200
CCBE1	83.8	95%	88%	Hennekam lymphangiectasia-lymphedema syndrome,235510
CD151	79.9	100%	96%	Nephropathy with pretibial epidermolysis bullosa and deafness,609057 [Blood group, Raph],179620
CDAN1	89.4	100%	97%	Dyserythropoietic anemia, congenital, type Ia,224120
CDH3	89.3	99%	95%	Ectodermal dysplasia,ectrodactyly and macular dystrophy,225280 Hypotrichosis, congenital, with juvenile macular dystrophy,601553
CDK4	120.9	96%	88%	{Melanoma, cutaneous malignant, 3},609048
CDKN2A	97.6	93%	93%	Melanoma and neural system tumor syndrome,155755 Pancreatic cancer/melanoma syndrome,606719 {Melanoma,cutaneous malignant, 2},155601
CDSN	10.9	49%	18%	Hypotrichosis 2,146520 Peeling skin syndrome 1,270300
CECR1	90.2	99%	96%	?Sneddon syndrome,182410 Polyarteritis nodosa, childhood-onset,615688
CERS3	81.1	100%	99%	Ichthyosis, congenital, autosomal recessive 9,615023
CHKB	77	91%	90%	Muscular dystrophy congenital megaconial type,602541
CHST14	118.4	100%	97%	Ehlers-Danlos syndrome musculocontractural type,601776
CHSY1	142.4	96%	94%	Temtamy preaxial brachydactyly syndrome,605282
CHUK	86.2	100%	97%	Cocoon syndrome,613630
CKAP2L	129.2	100%	100%	Filippi syndrome,272440
CLDN1	106.6	100%	100%	Ichthyosis,leukocyte vacuoles,alopecia and sclerosing cholangitis,607626
CNNM4	165.5	100%	96%	Jalili syndrome,217080
COL17A1	85.2	97%	92%	Epidermolysis bullosa,junctional,non-Herlitz type,226650
COL1A2	88.3	97%	91%	Ehlers-Danlos syndrome,cardiac valvular form,225320 Ehlers-Danlos syndrome, type VIIB,130060 Osteogenesis imperfecta, type II,166210 Osteogenesis imperfecta, type III,259420 Osteogenesis imperfecta, type IV,166220

COL3A1	67.6	97%	91%	Ehlers-Danlos syndrome,type III,130020 Ehlers-Danlos syndrome, type IV,130050
COL5A1	98.2	97%	96%	Ehlers-Danlos syndrome, classic type I,130000
COL5A2	82.1	98%	94%	Ehlers-Danlos syndrome, classic type I,130000
COL7A1	100.6	100%	99%	EBD inversa,226600 EBD, Bart type,132000 Epidermolysis bullosa dystrophica, AD,131750 Epidermolysis bullosa dystrophica, AR,226600 Epidermolysis bullosa pruriginosa,604129 Epidermolysis bullosa,pretibial,131850 Toenail dystrophy,isolated,607523
COX4I2	49.5	99%	90%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis,612714
COX7B	56.8	99%	98%	Linear skin defects with multiple congenital anomalies,300887
CPOX	79.2	100%	98%	Coproporphyria,121300 Harderoporphyria,121300
CSTA	101.2	100%	100%	Exfoliative ichthyosis,autosomal recessive,ichthyosis bullosa of Siemens-like,607936
CTC1	96.7	99%	96%	Cerebroretinal microangiopathy with calcifications and cysts,612199
CTSA	100.2	100%	99%	Galactosialidosis,256540
CTSC	95.5	100%	99%	Haim-Munk syndrome,245010 Papillon-Lefevre syndrome,245000 Periodontitis 1, juvenile,170650
CXCR4	193.2	100%	100%	Myelokathexis, isolated WHIM syndrome, 193670
CYLD	113.6	100%	99%	Brooke-Spiegler syndrome,605041 Cylindromatosis,familial,132700 Trichoepithelioma,multiple familial,1,601606
CYP26C1	55.3	100%	91%	Focal facial dermal dysplasia 4,614974
CYP4F22	91.1	99%	97%	Ichthyosis,congenital,autosomal recessive 5,604777
DCAF17	95.3	100%	95%	Woodhouse-Sakati syndrome,241080
DCLRE1C	97.2	90%	90%	Omenn syndrome,603554 Severe combined immunodeficiency, Athabascan type,602450
DDB2	90.8	100%	98%	Xeroderma pigmentosum,group E,DDB-negative subtype,278740
DHCR7	107	99%	98%	Smith-Lemli-Opitz syndrome,270400
DKC1	91.1	100%	98%	Dyskeratosis congenita X-linked,305000

DLX3	71.8	98%	93%	Amelogenesis imperfecta,type IV,104510 Trichodontoosseous syndrome,190320
DLX5	82.8	95%	88%	?Split-hand/foot malformation 1 with sensorineural hearing loss,220600
DOCK6	86.1	99%	94%	Adams-Oliver syndrome 2,614219
DOCK8	85.9	100%	98%	Hyper-IgE recurrent infection syndrome autosomal recessive,243700
DOLK	142.4	100%	100%	Congenital disorder of glycosylation, type Im,610768
DSC2	97.1	99%	97%	Arrhythmogenic right ventricular dysplasia 11Without/with mild palmoplantar keratoderma and woolly hair,610476
DSC3	95.6	99%	99%	?Hypotrichosis and recurrent skin vesicles,613102
DSE	91.9	92%	84%	?Ehlers-Danlos syndrome, musculocontractural type 2,615539
DSG1	147.8	100%	100%	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis and hyper IgE,615508 Keratosis palmoplantaris striata I,AD,148700
DSG3	146.1	99%	98%	No OMIM disease ID
DSG4	116.6	100%	100%	Hypotrichosis 6,607903
DSP	129.2	99%	98%	Arrhythmogenic right ventricular dysplasia 8,607450 Cardiomyopathy, dilated, with woolly hair and keratoderma,605676 Dilated cardiomyopathy with woolly hair, keratoderma and tooth agenesis,615821 Epidermolysis bullosa,lethal acantholytic,609638
DSPP	138.4	98%	96%	Deafness,autosomal dominant 36,with dentinogenesis,605594 Dentin dysplasia,type II,125420 Dentinogenesis imperfecta, Shields type II,125490 Dentinogenesis imperfecta, Shields type III, 125500
DTNBP1	107.6	100%	100%	Hermansky-Pudlak syndrome 7,614076 {Schizophrenia},181500
DUSP6	146.9	100%	96%	Hypogonadotropic hypogonadism 19 with or without anosmia,615269
EBP	95.6	99%	95%	Chondrodysplasia punctata X-linked dominant,302960
ECM1	115.3	100%	99%	Urbach-Wiethe disease,247100
EDA	89.3	99%	96%	Ectodermal dysplasia 1,hypohidrotic,X-linked,305100 Tooth agenesis,selective,X-linked 1,313500
EDAR	77.1	100%	98%	Ectodermal dysplasia 10A,hypohidrotic/hair/nail type, autosomal dominant,129490 Ectodermal dysplasia 10B,hypohidrotic/hair/tooth type, autosomal recessive,224900 [Hair morphology 1,hair thickness],612630
EDARADD	114.5	100%	98%	Ectodermal dysplasia 11A,hypohidrotic/hair/tooth type, autosomal dominant,614940 Ectodermal dysplasia 11B,hypohidrotic/hair/tooth type, autosomal recessive,614941

EDN3	91.5	100%	100%	Central hypoventilation syndrome congenital,209880 Waardenburg syndrome, type 4B,613265 {Hirshprung disease,susceptibility to,4},613712
EDNRA	113.1	100%	100%	mandibulofacial dysostosis with alopecia, 616367 {Migraine, resistance to},157300
EDNRB	137.1	100%	99%	ABCD syndrome,600501 Waardenburg syndrome, type 4A,277580 {Hirshprung disease, susceptibility to, 2},600155
EFEMP2	105.3	100%	100%	Cutis laxa,autosomal recessive,type IB,614437
EFNB1	106	100%	100%	Craniofrontonasal dysplasia,304110
EIF2AK3	106.5	92%	91%	Wolcott-Rallison syndrome,226980
ELN	68.3	100%	97%	Cutis laxa AD,123700 Supravalvar aortic stenosis,185500
ELOVL4	107.3	100%	100%	?Spinocerebellar ataxia 34,133190 Ichthyosis,spastic quadriplegia and mental retardation,614457 Stargardt disease 3,600110
ENAM	125.1	100%	100%	Amelogenesis imperfecta type IB,104500 Amelogenesis imperfecta type IC,204650
ENG	70.1	97%	86%	Telangiectasia,hereditary hemorrhagic,type 1,187300
ENPP1	106.7	96%	93%	Arterial calcification,generalized,of infancy 1,208000 Cole disease,615522 Hypophosphatemic rickets, autosomal recessive,2,613312 {Diabetes mellitus, non-insulin-dependent, susceptibility to},125853 {Obesity,susceptibility to},601665
EPG5	87.2	100%	99%	Vici syndrome,242840
ERCC2	87.2	99%	93%	Cerebrooculofacioskeletal syndrome 2,610756 Trichothiodystrophy 1, photosensitive,601675 Xeroderma pigmentosum, group D,278730
ERCC3	121.5	100%	100%	Trichothiodystrophy 2, photosensitive,616390 Xeroderma pigmentosum, group B,610651
ERCC4	140.8	99%	93%	Fanconi anemia,complementation group Q,615272 Xeroderma pigmentosum, group F,278760 Xeroderma pigmentosum, type F/Cockayne syndrome,278760 XFE progeroid syndrome,610965

ERCC5	113.8	98%	97%	Xeroderma pigmentosum, group G,278780 Xeroderma pigmentosum, group G/Cockayne syndrome,278780
ERCC6	141.3	98%	97%	Cerebrooculofacioskeletal syndrome 1,214150 Cockayne syndrome, type B,133540 De Sanctis-Cacchione syndrome,278800 UV-sensitive syndrome I,600630 {Lung cancer, susceptibility to},211980 {Macular degeneration, age-related, susceptibility to 5},613761
ERCC8	88.4	100%	98%	Cockayne syndrome type A,216400 UV-sensitive syndrome 2,614621
EVC	73.6	92%	88%	Ellis-van Creveld syndrome,225500 Weyers acrodental dysostosis,193530
EVC2	100.1	94%	92%	Ellis-van Creveld syndrome,225500 Weyers acrodental dysostosis,193530
EXPH5	143	99%	99%	Epidermolysis bullosa,nonspecific,autosomal recessive,615028
FAM111B	168.9	100%	99%	Poikiloderma, fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis,615704
FAM20A	68.4	99%	85%	Amelogenesis imperfecta,type IG (enamel-renal syndrome),204690
FAM20C	77.6	92%	87%	Raine syndrome,259775
FAM83H	73.2	99%	97%	Amelogenesis imperfecta type 3,130900
FANCA	87.6	99%	97%	Fanconi anemia complementation group A,227650
FANCB	121.9	100%	97%	Fanconi anemia complementation group B,300514
FANCC	76.9	100%	94%	Fanconi anemia complementation group C,227645
FANCD2	92.2	87%	86%	Fanconi anemia complementation group D2,227646
FANCE	86.4	97%	92%	Fanconi anemia complementation group E,600901
FANCF	143.4	100%	100%	Fanconi anemia complementation group F,603467
FANCG	120.8	99%	96%	Fanconi anemia complementation group G,614082
FANCI	118.1	100%	100%	Fanconi anemia complementation group I,609053
FANCL	93.8	100%	100%	Fanconi anemia complementation group L,614083
FANCM	117.3	100%	99%	Fanconi anemia complementation group M,614087
FAT4	159.5	100%	100%	Hennekam lymphangiectasia-lymphedema syndrome 2,616006 Van Maldergem syndrome 2,615546
FBLN5	72	91%	89%	Cutis laxa,autosomal dominant 2,614434 Cutis laxa,autosomal recessive,type IA,219100 Macular degeneration,age-related,3,608895

FDPS	47.4	85%	69%	Porokeratosis 9,multiple types,616631
FECH	104.2	100%	100%	Protoporphyria erythropoietic autosomal recessive,177000
FERMT1	101.6	100%	100%	Kindler syndrome,173650
FGF10	110.7	100%	100%	Aplasia of lacrimal and salivary glands,180920 LADD syndrome,149730
FGF23	82.6	96%	91%	Hypophosphatemic rickets,autosomal dominant,193100 Osteomalacia,tumor-induced Tumoral calcinosis,hyperphosphatemic,familial,211900
FGF3	88.2	100%	98%	Deafness,congenital with inner ear agenesis,microtia and microdontia,610706
FGF5	174.1	100%	100%	trichomegaly,190330
FGF8	52.7	84%	68%	Hypogonadotropic hypogonadism 6 with or without anosmia,612702
FGFR1	113.7	99%	97%	Hartsfield syndrome,615465 Hypogonadotropic hypogonadism 2 with or without anosmia,147950 Jackson-Weiss syndrome,123150 Osteoglophonic dysplasia,166250 Pfeiffer syndrome,101600 Trigonocephaly 1,190440
FGFR2	115.3	97%	97%	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis,207410 Apert syndrome,101200 Beare-Stevenson cutis gyrata syndrome,123790 Bent bone dysplasia syndrome,614592 Craniofacial-skeletal-dermatologic dysplasia,101600
FGFR3	71.9	95%	87%	Achondroplasia,100800 Bladder cancer,somatic,109800 CATSHL syndrome,610474 Cervical cancer,somatic,603956 Colorectal cancer,somatic,114500 Crouzon syndrome with acanthosis nigricans,612247 Hypochondroplasia,146000 LADD syndrome,149730
FH	85.3	96%	89%	Fumarase deficiency,606812 Leiomyomatosis and renal cell cancer,150800
FKBP10	81.8	100%	99%	Bruck syndrome 1,259450 Osteogenesis imperfecta type XI,610968
FKBP14	124.6	100%	100%	Ehlers-Danlos syndrome with progressive kyphoscoliosis myopathy and hearing loss,614557

FLCN	110.5	100%	97%	Birt-Hogg-Dube syndrome,135150 Colorectal cancer,somatic,114500 Pneumothorax,primary spontaneous,173600 Renal carcinoma,chromphobe,somatic,144700
FLG	44.1	98%	81%	Ichthyosis vulgaris,146700 {Dermatitis,atopic,susceptibility to,2},605803
FLG2	239.9	100%	100%	No OMIM disease ID
FLT4	91.5	99%	98%	Hemangioma,capillary infantile,somatic,602089 Lymphedema,hereditary,IA,153100
FNIP1	149.3	100%	99%	Familial multiple discoid fibromas,190340
FOXC2	83.3	100%	98%	Lymphedema-distichiasis syndrome with/without renal disease and diabetes mellitus,153400
FOXE1	40.3	100%	84%	Bamforth-Lazarus syndrome,241850
FOXN1	113.5	100%	96%	T-cell immunodeficiency congenital alopecia and nail dystrophy,601705
FOXP3	84.9	99%	94%	Immunodysregulation,polyendocrinopathy and enteropathy,X-linked,304790 {Diabetes mellitus,type I,susceptibility to},222100
FREM1	107.3	99%	99%	Bifid nose with or without anorectal and renal anomalies,608980 Manitoba oculotrichoanal syndrome,248450 Trigonocephaly 2,614485
FUCA1	80.4	100%	99%	Fucosidosis,230000
FZD6	144.5	100%	100%	Nail disorder,nonsyndromic,congenital 10 (claw-shaped nails),614157
GALNS	68.5	93%	92%	Mucopolysaccharidosis IVA,253000
GALNT3	106.8	100%	100%	Tumoral calcinosis,hyperphosphatemic,familial,211900
GAN	128.2	100%	99%	Giant axonal neuropathy-1,256850
GATA2	105	96%	91%	Emberger syndrome,614038 Immunodeficiency 21,614172 {Leukemia, acute myeloid, susceptibility to},601626 {Myelodysplastic syndrome, susceptibility to},614286
GDF2	135.9	100%	100%	Telangiectasia, hereditary hemorrhagic, type 5,615506

GDF5	96	100%	100%	Acromesomelic dysplasia,Hunter-Thompson type,201250 Brachydactyly,type A1,C,615072 Brachydactyly,type A2,112600 Brachydactyly,type C,113100 Chondrodysplasia,Grebe type,200700 Du Pan syndrome,228900 Multiple synostoses syndrome 2,610017 Symphalangis
GGCX	93.9	100%	97%	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency,610842 Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450
GJA1	66	85%	73%	Atrioventricular septal defect 3,600309 Craniometaphyseal dysplasia, autosomal recessive,218400 Erythrokeratodermia variabilis et progressiva,133200 Hypoplastic left heart syndrome 1,241550 Oculodentodigital dysplasia,164200 Oculodentodigital dysplas
GJB2	174.6	100%	100%	Bart-Pumphrey syndrome,149200 Deafness,autosomal dominant 3A,601544 Deafness,autosomal recessive 1A,220290 Hystrix-like ichtyosis-deafness syndrome,602540 Keratitis-ichtyosis-deafness syndrome,148210 Keratoderma,palmoplantar,with deafness,148350
GJB3	128.3	100%	100%	Deafness autosomal dominant 2B,612644 Deafness,digenic,GJB2/GJB3,220290 Erythrokeratodermia variabilis et progressiva,133200
GJB4	137.7	100%	100%	Erythrokeratodermia variabilis with erythema gyratum repens,133200
GJB6	141.9	100%	100%	Deafness,autosomal dominant 3B,612643 Deafness,autosomal recessive 1B,612645 Deafness,digenic GJB2/GJB6,220290 Ectodermal dysplasia 2,Clouston type,129500
GJC2	52.1	92%	82%	Leukodystrophy,hypomyelinating 2,608804 Lymphedema, hereditary,IC,613480 Spastic paraplegia 44,autosomal recessive,613206
GLA	92.4	100%	98%	Fabry disease,301500

GLB1	79	98%	95%	GM1-gangliosidosis type I,230500 GM1-gangliosidosis type II,230600 GM1-gangliosidosis type III,230650 Mucopolysaccharidosis type IVB (Morquio),253010
GLMN	88.1	99%	96%	Glomuvenous malformations,138000
GMPPA	125.8	100%	100%	Alacrima, achalasia, and mental retardation syndrome ,615510
GNA11	86.6	100%	99%	Hypocalcemia,autosomal dominant 2,615361 Hypocalciuric hypercalcemia,type II,145981
GNAQ	63.2	96%	93%	Capillary malformations,congenital,1, somatic,mosaic,163000 Sturge-Weber syndrome, somatic, mosaic,185300
GNAS	125.5	100%	97%	Acromegaly, somatic,102200 ACTH-independent macronodular adrenal hyperplasia,219080 McCune-Albright syndrome,somatic,mosaic,174800 Osseous heteroplasia,progressive,166350 Pseudohypoparathyroidism Ia,103580 Pseudohypoparathyroidism Ib,603233
GORAB	154.2	100%	98%	Geroderma osteodysplasticum,231070
GPR143	48.7	90%	80%	Nystagmus 6,congenital,X-linked,300814 Ocular albinism, type I, Nettleship-Falls type,300500
GRHL2	102	100%	99%	Deafness,autosomal dominant 28,608641 Ectodermal dysplasia/short stature syndrome,616029
GRHL3	101.2	100%	100%	Van der Woude syndrome 2, 606713
GSN	76.6	97%	91%	Amyloidosis Finnish type,105120
GTF2H5	89.9	100%	100%	Trichothiodystrophy 3,photosensitive,616395
HCCS	107.1	100%	99%	Linear skin defects with multiple congenital anomalies,309801
HDAC8	98.2	100%	99%	Cornelia de Lange syndrome 5,300882 Wilson-Turner syndrome,309585
HERC2	62.1	63%	59%	Mental retardation, autosomal recessive 38,615516 [Skin/hair/eye pigmentation 1],227220
HLCS	140.4	100%	100%	Holocarboxylase synthetase deficiency,253270
HMBS	91.4	100%	96%	Porphyria acute intermittent,176000
HMGB3	34	98%	50%	?Microphthalmia, syndromic 13,300915
HOXC13	83.2	100%	98%	Ectodermal dysplasia 9 hair/nail type,614931
HPS1	73.1	99%	90%	Hermansky-Pudlak syndrome 1,203300

HPS3	109.9	100%	98%	Hermansky-Pudlak syndrome 3,614072
HPS4	109.7	100%	98%	Hermansky-Pudlak syndrome 4,614073
HPS5	93.6	96%	95%	Hermansky-Pudlak syndrome 5,614074
HPS6	88.1	100%	82%	Hermansky-Pudlak syndrome 6,614075
HR	75.4	99%	93%	Alopecia universalis,203655 Atrichia with papular lesions,209500 Hypotrichosis 4,146550
HRAS	90.1	100%	100%	Congenital myopathy with excess of muscle spindles,218040 Costello syndrome,218040 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic,163200 {Bladder cancer, somatic},109800 {Nevus sebaceous or woolly hair nevus, somatic},162900
HTRA1	72.3	85%	80%	CARASIL syndrome,600142 {Macular degeneration,age-related,7},610149 {Macular degeneration,age-related,neovascular type},610149
HYAL1	84.4	98%	93%	Mucopolysaccharidosis type IX,601492
IDUA	77.9	95%	90%	Mucopolysaccharidosis Ih,607014 Mucopolysaccharidosis Ih/s,607015 Mucopolysaccharidosis Is,607016
IFT122	78.2	96%	95%	Cranioectodermal dysplasia 1,218330
IFT43	86.1	100%	100%	Cranioectodermal dysplasia 3,614099
IKBKG	25.9	26%	26%	Ectodermal dysplasia,hypohidrotic with immune deficiency,300291 Ectodermal dysplasia,anhydrotic,lymphedema and immunodeficiency,300301 Immunodeficiency 33,300636 Immunodeficiency,isolated,300584 Incontinentia pigmenti,308300
IL17RA	87.2	98%	89%	?Candidiasis,familial 5,autosomal recessive,613953
IL17RD	100.3	100%	97%	Hypogonadotropic hypogonadism 18 with or without anosmia,615267
IL1RN	110.9	100%	100%	Interleukin 1 receptor antagonist deficiency,612852 {Gastric cancer risk after H.pylori infection},137215 {Microvascular complications of diabetes 4},612628
IL31RA	127.7	100%	97%	Amyloidosis,primary localized cutaneous 2,613955
IL36RN	89	100%	100%	Psoriasis 14, pustular,614204

INSR	120.5	98%	95%	Diabetes mellitus,insulin-resistant,with acanthosis nigricans,610549 Hyperinsulinemic hypoglycemia,familial,5,609968 Leprechaunism,246200 Rabson-Mendenhall syndrome,262190
IRF4	112.9	100%	99%	Multiple myeloma,254500 [Skin/hair/eye pigmentation, variation in,8],611724
IRF6	91	96%	93%	Orofacial cleft 6,608864 Popliteal pterygium syndrome 1,119500 van der Woude syndrome,119300
ITGA3	107.3	100%	92%	Interstitial lung disease, nephrotic syndrome and epidermolysis bullosa, congenital,614748
ITGA6	123.5	99%	98%	Epidermolysis bullosa,junctional, with pyloric stenosis,226730
ITGB4	79.9	97%	94%	Epidermolysis bullosa of hands and feet,131800 Epidermolysis bullosa,junctional,non-Herlitz type,226650 Epidermolysis bullosa,junctional,with pyloric atresia,226730
ITGB6	101.7	97%	95%	Amelogenesis imperfecta, type IH,616221
JUP	72.2	98%	93%	Arrhythmogenic right ventricular dysplasia 12,611528 Naxos disease,601214
KAL1	78.4	96%	91%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1),308700
KANK2	110.3	100%	100%	palmoplantar keratoderma and woolly hair,616099
KAT6B	140.2	100%	100%	Genitopatellar syndrome,606170 SBBYSS syndrome,603736
KCNH1	110	100%	99%	Temple-Baraitser syndrome,611816 Zimmermann-Laband syndrome,135500
KCNK9	127.2	100%	100%	Birk-Barel mental retardation dysmorphism syndrome,612292
KIF11	95.9	100%	98%	Microcephaly with or without chorioretinopathy lymphedema or mental retardation,152950
KIT	106.6	98%	96%	Gastrointestinal stromal tumor,familial,606764 Germ cell tumors,273300 Leukemia,acute myeloid,601626 Mast cell disease,154800 Piebaldism,172800
KITLG	74.7	100%	98%	Hyperpigmentation familial progressive 2,145250 [Skin/hair/eye pigmentation 7],611664
KLK4	143.3	100%	100%	Amelogenesis imperfecta type IIA1,204700
KLLN	109.6	100%	100%	Cowden syndrome 4,615107

KMT2D	102	99%	98%	Kabuki syndrome 1,147920
KRAS	65.1	95%	89%	Bladder cancer,somatic,109800 Breast cancer,somatic,114480 Cardiofaciocutaneous syndrome 2,615278 Gastric cancer,somatic,137215 Lung cancer,somatic,211980 Noonan syndrome 3,609942 Pancreatic carcinoma,somatic,260350 SFM syndrome,somatic mosaic,1632
KRT1	100	100%	98%	Epidermolytic hyperkeratosis,113800 Ichthyosis histrix,Curth-Macklin type,146590 Ichthyosis, cyclic, with epidermolytic hyperkeratosis,607602 Keratosis palmoplantaris striata III,607654 Palmoplantar keratoderma,epidermolytic,144200
KRT10	95.3	97%	88%	Epidermolytic hyperkeratosis,113800 Ichthyosis with confetti,609165 Ichthyosis,cyclic,with epidermolytic hyperkeratosis,607602
KRT13	91.2	100%	99%	White sponge nevus 2,615785
KRT14	25.9	70%	50%	Dermatopathia pigmentosa reticularis,125595 Epidermolysis bullosa simplex,Dowling-Meara type,131760 Epidermolysis bullosa simplex,Koebner type,131900 Epidermolysis bullosa simplex,recessive 1,601001 Epidermolysis bullosa simplex,Weber-Cockayne type,13
KRT16	7.5	24%	3%	Pachyonychia congenita 1,167200 Palmoplantar keratoderma,nonepidermolytic,focal,613000
KRT17	11.2	48%	15%	Pachyonychia congenita 2,167210 Steatocystoma multiplex,184500
KRT2	112.4	99%	97%	Ichthyosis bullosa of Siemens,146800
KRT4	83.8	100%	99%	White sponge nevus 1,193900
KRT5	73.4	97%	92%	Dowling-Degos disease 1,179850 Epidermolysis bullosa simplex,Dowling-Meara type,131760 Epidermolysis bullosa simplex,Koebner type,131900 Epidermolysis bullosa simplex,recessive 1,601001 Epidermolysis bullosa simplex,Weber-Cockayne type,131800

KRT6A	32.6	68%	45%	Pachyonychia congenita 3,167200
KRT6B	32.2	76%	52%	Pachyonychia congenita Jackson-Lawler type,615726
KRT6C	22	52%	36%	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse,615735
KRT71	106.8	99%	93%	Hypotrichosis 13,615896
KRT74	81.2	93%	86%	?Ectodermal dysplasia 7, hair/nail type,614929 ?Hypotrichosis 3,613981 Woolly hair, autosomal dominant,194300
KRT75	94.5	100%	94%	{Pseudofolliculitis barbae,susceptibility to},612318
KRT81	19.9	61%	37%	Monilethrix,158000
KRT83	23.6	63%	39%	Monilethrix,158000
KRT85	34.9	79%	58%	Ectodermal dysplasia 4 hair/nail type,602032
KRT86	24.2	59%	45%	Monilethrix,158000
KRT9	106.7	98%	94%	Epidermolytic palmoplantar keratoderma,144200
LAMA3	97.1	99%	98%	Epidermolysis bullosa,generalized atrophic benign,226650 Epidermolysis bullosa,junctional,Herlitz type,226700 Laryngoonychocutaneous syndrome,245660
LAMB3	72.5	99%	95%	Amelogenesis imperfecta,type IA,104530 Epidermolysis bullosa,junctional,Herlitz type,226700 Epidermolysis bullosa,junctional,non-Herlitz type,226650
LAMC2	101.9	99%	97%	Epidermolysis bullosa,junctional,Herlitz type,226700 Epidermolysis bullosa,junctional,non-Herlitz type,226650
LAMTOR2	73.4	100%	100%	Immunodeficiency due to defect in MAPBP-interacting protein,610798
LDHA	41.8	70%	63%	Glycogen storage disease XI,612933
LDLRAP1	82.9	96%	90%	Hypercholesterolemia,familial,autosomal recessive,603813
LEMD3	109.8	100%	99%	Buschke-Ollendorff syndrome,166700 Melorheostosis with osteopoikilosis,155950 Osteopoikilosis,166700
LIPH	118	100%	98%	Hypotrichosis 7,604379 Woolly hair,autosomal recessive 2,with or without hypotrichosis
LIPN	108.4	100%	99%	Ichthyosis,congenital,autosomal recessive 8,613943
LMBRD1	111.9	100%	100%	Methylmalonic aciduria and homocystinuria cblF type,277380

LMNA	69.9	98%	88%	Cardiomyopathy dilated 1A,115200 Charcot-Marie-Tooth disease,type 2B1,605588 Emery-Dreifuss muscular dystrophy 2, AD,181350 Emery-Dreifuss muscular dystrophy 3,AR,616516 Heart-hand syndrome,Slovenian type,610140 Hutchinson-Gilford progeria,176670
LMX1B	82.4	98%	94%	Nail-patella syndrome,161200
LONP1	81	97%	94%	CODAS syndrome,600373
LOR	37.3	99%	85%	Vohwinkel syndrome with ichthyosis,604117
LPAR6	114.5	100%	100%	Hypotrichosis 8,278150 Woolly hair,autosomal recessive 1,with or without hypotrichosis,278150
LPIN2	75.8	99%	95%	Majeed syndrome,609628
LTBP3	74.5	99%	95%	Dental anomalies and short stature,601216
LTBP4	83	97%	89%	Cutis laxa autosomal recessive type IC,613177
LYST	116.3	99%	97%	Chediak-Higashi syndrome,214500
LYZ	101	100%	100%	Amyloidosis,renal,105200
MAP2K1	92.8	96%	81%	Cardiofaciocutaneous syndrome 3,615279
MAP2K2	111.8	100%	98%	Cardiofaciocutaneous syndrome 4,615280
MBTPS2	126.7	100%	100%	?Olmsted syndrome,X-linked,300918 IFAP syndrome with or without BRESHECK syndrome,308205 Keratosis follicularis spinulosa decalvans,X-linked,308800
MED12	122.4	97%	94%	Lujan-Fryns syndrome,309520 Ohdo syndrome,X-linked,300895 Opitz-Kaveggia syndrome,305450
MEFV	113.7	96%	95%	Familial Mediterranean fever AD,134610 Familial Mediterranean fever AR,249100
MGP	71.7	100%	99%	Keutel syndrome,245150
MITF	136.6	100%	100%	Tietz albinism-deafness syndrome,103500 Waardenburg syndrome, type 2A,193510 Waardenburg syndrome/ocular albinism, digenic,103470 {Melanoma,cutaneous malignant,susceptibility to 8},614456
MLH1	98.7	100%	99%	Colorectal cancer,hereditary,nonpolyposis type 2,609310 Mismatch repair cancer syndrome,276300 Muir-Torre syndrome,158320

MLPH	77.3	93%	88%	Griscelli syndrome type 3,609227
MMACHC	175.4	100%	100%	Methylmalonic aciduria and homocystinuria cblC type,277400
MMP2	98.9	100%	99%	Torg-Winchester syndrome,259600
MMP20	94	100%	98%	Amelogenesis imperfecta type IIA2,612529
MPLKIP	66	100%	100%	Trichothiodystrophy nonphotosensitive 1,234050
MRE11A	90.5	99%	99%	Ataxia-telangiectasia-like disorder,604391
MSH2	98.8	98%	97%	Colorectal cancer,hereditary,nonpolyposis type 1,120435 Mismatch repair cancer syndrome,276300 Muir-Torre syndrome,158320
MSX1	55.6	100%	82%	Ectodermal dysplasia 3,Witkop type,189500 Orofacial cleft 5,608874 Tooth agenesis,selective,1,with or without orofacial cleft,106600
MTOR	98.6	100%	98%	Smith-Kingsmore syndrome,616638
MUTYH	110.4	100%	99%	Adenomas,multiple colorectal,608456 Colorectal denomatous polyposis,autosomal recessive,with pilomatricomas,132600 Gastric cancer,somatic,613659
MVD	68.6	100%	98%	Porokeratosis 7,multiple types,614714
MVK	86.1	100%	99%	Hyper-IgD syndrome,260920 Mevalonic aciduria,610377 Porokeratosis 3,disseminated superficial actinic,175900
MYH8	101.5	97%	88%	Carney complex variant,608837 Trismus-pseudocamptodactyly syndrome,158300
MYO5A	91.4	99%	96%	Griscelli syndrome type 1,214450
NAA10	94.2	97%	97%	N-terminal acetyltransferase deficiency,300855 ?Microphthalmia,syndromic 1,309800
NAGA	82.1	100%	95%	Kanzaki disease,609242 Schindler disease,609241
NBAS	97.1	100%	98%	Short stature,optic nerve atrophy and Pelger-Huet anomaly,614800 Infantile liver failure syndrome 2,616483
NCSTN	79.2	97%	92%	Acne inversa familial 1,142690
NDUFB11	95.4	99%	99%	Linear skin defects with multiple congenital anomalies 3,300952
NF1	78.6	82%	81%	Neurofibromatosis, type 1,162200
NFKBIA	95.9	100%	99%	Ectodermal dysplasia anhidrotic with T-cell immunodeficiency,612132
NHP2	47	100%	93%	Dyskeratosis congenita, autosomal recessive 2,613987

NIPAL4	114.1	100%	98%	Ichthyosis,congenital,autosomal recessive 6,612281
NIPBL	113.4	99%	98%	Cornelia de Lange syndrome 1,122470
NLRP1	105.9	99%	98%	?Corneal intraepithelial dyskeratosis and ectodermal dysplasia,615225 {Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579
NLRP3	116.6	100%	99%	CINCA syndrome,607115 Familial cold-induced inflammatory syndrome 1,120100 Muckle-Wells syndrome,191900
NME1	129.5	100%	100%	Neuroblastoma,256700
NOD2	91.7	100%	98%	Blau syndrome,186580 Sarcoidosis,early-onset,609464 {Inflammatory bowel disease 1},266600 {Psoriatic arthritis,susceptibility to},607507
NOP10	169.5	100%	100%	Dyskeratosis congenita, autosomal recessive 1,224230
NOTCH1	69.6	98%	87%	Aortic valve disease,109730 Adams-Oliver syndrome 5,616028
NRAS	135	100%	100%	Autoimmune lymphoproliferative syndrome type IV,614470 Colorectal cancer,somatic,114500 Epidermal nevus,somatic,162900 Melanocytic nevus syndrome,congenital,somatic,137550 Neurocutaneous melanosis,somatic,249400 Noonan syndrome 6,613224
NSD1	119.6	100%	99%	Beckwith-Wiedemann syndrome,130650 Leukemia,acute myeloid,601626 Sotos syndrome,117550
NSDHL	100	99%	97%	CHILD syndrome,308050 CK syndrome,300831
OCA2	89.6	100%	98%	Albinism brown oculocutaneous,203200 [Skin/hair/eye pigmentation 1],227220
ODAM	101.3	100%	100%	No OMIM disease ID
OFD1	68.6	93%	88%	?Retinitis pigmentosa 23,300424 Joubert syndrome 10,300804 Orofaciodigital syndrome 1,311200 Simpson-Golabi-Behmel syndrome, type 2,300209
OSMR	134.1	100%	100%	Amyloidosis primary localized cutaneous 1,105250

PAH	79.8	96%	92%	Phenylketonuria,261600
PALB2	122.1	99%	97%	Fanconi anemia complementation group N,610832 {Breast cancer,susceptibility to},114480 {Pancreatic cancer,susceptibility to 3},613348
PAX3	107.4	99%	97%	Craniofacial-deafness-hand syndrome,122880 Rhabdomyosarcoma 2,alveolar,268220 Waardenburg syndrome,type 1,193500 Waardenburg syndrome,type 3,148820
PAX9	209.9	99%	99%	Tooth agenesis selective 3,604625
PCNA	87.3	100%	100%	Ataxia-telangiectasia-like disorder 2,615919
PDGFB	66.9	100%	97%	Dermatofibrosarcoma protuberans,607907 Basal ganglia calcification,idiopathic,5,615483 Meningioma, SIS-related,607174
PDGFRB	82.4	100%	98%	Basal ganglia calcification idiopathic 4,615007 Myeloproliferative disorder with eosinophilia, 131440 Myofibromatosis, infantile, 1, 228550
PEPD	63.9	100%	90%	Prolidase deficiency,170100
PEX7	90.4	90%	81%	Peroxisome biogenesis disorder 9B,614879 Chondrodysplasia punctata, rhizomelic, type 1,215100
PHEX	118.8	98%	98%	Hypophosphatemic rickets X-linked dominant,307800
PHGDH	86.3	100%	99%	Phosphoglycerate dehydrogenase deficiency,601815 Neu-Laxova syndrome 1,256520
PHYH	84.3	100%	98%	Refsum disease,266500
PIEZO1	85.8	98%	94%	Dehydrated hereditary stomatocytosis,pseudohyperkalemia and/or perinatal edema ,194380
PIGA	134.6	100%	99%	Multiple congenital anomalies-hypotonia-seizures syndrome 2,300868 Paroxysmal nocturnal hemoglobinuria,somatic,300818
PIGN	102.3	100%	99%	Multiple congenital anomalies-hypotonia-seizures syndrome 1,614080
PIGV	165	100%	100%	Hyperphosphatasia with mental retardation syndrome 1,239300

PIK3CA	112.1	93%	91%	Breast cancer somatic,114480 CLOVE syndrome, somatic,612918 Colorectal cancer,somatic,114500 Cowden syndrome 5,615108 Gastric cancer,somatic,613659 Hepatocellular carcinoma,somatic,114550 Keratosis,seborrheic,somatic,182000 Megalencephaly-capillar
PITX2	116.6	95%	89%	Axenfeld-Rieger syndrome type 1,180500 Iridogoniodysgenesis,type 2,137600 Peters anomaly,604229 Ring dermoid of cornea,180550
PKP1	86.1	97%	89%	Ectodermal dysplasia/skin fragility syndrome,604536
PLCD1	105.5	99%	95%	Nail disorder nonsyndromic congenital 3 (leukonychia),151600
PLCG2	105.4	100%	99%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome,614878 Familial cold autoinflammatory syndrome 3, 614468
PLEC	97.2	100%	96%	?Epidermolysis bullosa simplex with nail dystrophy,616487 Epidermolysis bullosa simples with muscular dystrophy,226670 Epidermolysis bullosa simplex with pyloric atresia,612138 Epidermolysis bullosa simplex, Ogna type, 131950 Muscular dystrophy,limb-g
PLG	64.4	75%	68%	Dysplasminogenemia,217090
PLIN1	44.2	92%	67%	Lipodystrophy familial partial type 4,613877
PLOD1	78.1	100%	97%	Ehlers-Danlos syndrome type VI,225400
PMS2	68.3	56%	55%	Colorectal cancer hereditary nonpolyposis type 4,614337 Mismatch repair cancer syndrome,276300
PMVK	79.1	97%	91%	Porokeratosis 1,multiple types,175800
PNPLA1	121.9	100%	100%	Ichthyosis congenital autosomal recessive 10,615024
PNPLA2	76.4	97%	93%	Neutral lipid storage disease with myopathy,610717
POC1A	101.6	98%	94%	Short stature onychodysplasia facial dysmorphism and hypotrichosis,614813
POFUT1	110	100%	98%	Dowling-Degos disease 2,615327
POGLUT1	103.1	98%	96%	Dowling-Degos disease 4,615696
POLD1	78.3	94%	91%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome,615381 {Colorectal cancer,susceptibility to,10},612591

POLH	141	98%	96%	Xeroderma pigmentosum variant type,278750
POLR1C	105.5	90%	86%	Treacher Collins syndrome 3,248390 Leukodystrophy, hypomyelinating,11,616494
POLR1D	167.4	100%	100%	Treacher Collins syndrome 2,613717
POLR3A	85.6	99%	95%	Leukodystrophy hypomyelinating 7 with or without oligodontia and/or hypogonadotropic hypogonadism,607694
POLR3B	100.3	99%	98%	Leukodystrophy hypomyelinating 8 with or without oligodontia and/or hypogonadotropic hypogonadism,614381
POMC	49.1	86%	75%	Obesity adrenal insufficiency and red hair due to POMC deficiency,609734 {Obesity,early-onset,susceptibility to},601665
POMP	153.1	100%	100%	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma,601952
PORCN	113.4	92%	90%	Focal dermal hypoplasia,305600
PPOX	101.1	100%	97%	Porphyria variegata,176200
PQBP1	121.6	100%	99%	Renpenning syndrome,309500
PRKAR1A	94.7	91%	83%	Acrodysostosis 1 with or without hormone resistance,101800 Carney complex,type 1,160980 Myxoma,intracardiac,255960 Pigmented nodular adrenocortical disease,primary,1,610489
PSEN1	92.5	100%	96%	Acne inversa familial 3,613737 Alzheimer disease,type 3,607822 Cardiomyopathy,dilated,1U,613694 Dementia,frontotemporal,600274 Pick disease,172700
PSENEN	117.2	100%	100%	Acne inversa familial 2,613736
PSMB8	10	38%	7%	Autoinflammation lipodystrophy and dermatosis syndrome,256040
PSTPIP1	57	96%	89%	Pyogenic sterile arthritis pyoderma gangrenosum and acne,604416
PTCH1	86.9	99%	95%	Basal cell carcinoma somatic,605462 Basal cell nevus syndrome, 109400 Holoprosencephaly-7,610828
PTCH2	81.1	99%	96%	Basal cell carcinoma somatic,605462 Basal cell nevus syndrome,109400 Medulloblastoma,155255
PTDSS1	112.6	100%	100%	Lenz-Majewski hyperostotic dwarfism,151050

PTEN	122.4	99%	94%	Bannayan-Riley-Ruvalcaba syndrome,153480 Cowden syndrome 1,158350 Endometrial carcinoma,somatic,608089 Macrocephaly/autism syndrome,605309 Malignant melanoma,somatic,155600 Squamous cell carcinoma,head and neck,somatic,275355
PTHLH	146.7	100%	100%	Brachydactyly type E2,613382 Humoral hypercalcemia of malignancy
PTPN11	41.7	83%	68%	LEOPARD syndrome 1,151100 Leukemia,juvenile myelomonocytic,607785 Metachondromatosis,156250 Noonan syndrome 1,163950
PTPN14	118.4	100%	99%	Choanal atresia and lymphedema,613611
PTPRF	96.4	99%	97%	Breasts and/or nipples, aplasia or hypoplasia of, 2,616001
PTRF	146.8	100%	100%	Lipodystrophy congenital generalized type 4,613327
PVRL1	76.5	100%	99%	Cleft lip/palate-ectodermal dysplasia syndrome,225060 Orofacial cleft 7,225060
PVRL4	90.5	100%	96%	Ectodermal dysplasia-syndactyly syndrome 1,613573
PYCR1	82	100%	98%	Cutis laxa autosomal recessive type IIB,612940 Cutis laxa autosomal recessive type IIIB,614438
RAB23	127.8	100%	100%	Carpenter syndrome,201000
RAB27A	119.7	100%	100%	Griscelli syndrome type 2,607624
RAD21	86.4	98%	90%	Cornelia de Lange syndrome 4,614701
RAD50	113.2	100%	99%	Nijmegen breakage syndrome-like disorder,613078
RAF1	89.3	100%	98%	LEOPARD syndrome 2,611554 Cardiomyopathy,dilated,1NN,615916 Noonan syndrome 5,611553
RAG1	132.2	100%	100%	Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection and autoimmunity,609889 Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554
RAG2	187.4	100%	100%	Combined cellular and humoral immune defects with granulomas,233650 Omenn syndrome,603554 Severe combined immunodeficiency,B cell-negative,601457

RAI1	135	99%	98%	Smith-Magenis syndrome,182290
RBBP8	110.5	100%	100%	Jawad syndrome,251255 Pancreatic carcinoma,somatic Seckel syndrome 2,606744
RBM28	104.4	100%	98%	Alopecia neurologic defects and endocrinopathy syndrome,612079
RBP4	68	92%	86%	Retinol dystrophy iris coloboma and comedogenic acne syndrome,615147 Microphthalmia,isolated,with coloboma 10,616428
RBPJ	70.6	99%	94%	Adams-Oliver syndrome 3,614814
RECQL4	91.5	98%	96%	Baller-Gerold syndrome,218600 RAPADILINO syndrome,266280 Rothmund-Thomson syndrome,268400
RHBDF2	62.5	96%	90%	Tylosis with esophageal cancer,148500
RIN2	100.4	99%	97%	Macrocephaly alopecia cutis laxa and scoliosis,613075
RIPK4	101.1	100%	96%	Popliteal pterygium syndrome 2 lethal type,263650
RNASEH2A	96.1	99%	94%	Aicardi-Goutieres syndrome 4,610333
RNASEH2B	102.3	99%	97%	Aicardi-Goutieres syndrome 2,610181
RNASEH2C	133.2	100%	100%	Aicardi-Goutieres syndrome 3,610329
ROGDI	94.2	97%	95%	Kohlschutter-Tonz syndrome,226750
RPL21	50.4	98%	81%	Hypotrichosis 12,615885
RSPO1	45.8	88%	81%	Palmoplantar hyperkeratosis and true hermaphroditism,610644 Palmoplantar hyperkeratosis with squamous cell skin carcinoma and sex revearsal,610644
RSPO4	82.7	100%	100%	Anonychia congenita,206800
RTEL1	73.3	99%	92%	Dyskeratosis congenita,autosomal recessive 5,615190 Dyskeratosis congenita,autosomal dominant 4,615190 Pulmonary fibrosis and/or bone marrow failure,telomere-related,616373
RUNX2	93.6	74%	74%	Cleidocranial dysplasia,119600 Cleidocranial dysplasia, forme fruste,dental anomalies only,119600 Cleidocranial dysplasia, forme fruste,with brachydactyly,119600 Metaphyseal dysplasia with maxillary hypoplasia with/without brachydactyly,156510
SAMD9	178.2	100%	100%	Tumoral calcinosis familial normophosphatemic,610455
SAMHD1	112.6	100%	98%	Aicardi-Goutieres syndrome 5,612952 Chilblain lupus 2,614415
SART3	86.6	100%	96%	No OMIM disease ID
SAT1	145.7	100%	100%	Keratosis follicularis spinulosa decalvans,308800

SATB2	100.2	100%	96%	Cleft palate and mental retardation,119540
SCN10A	117.2	99%	98%	Epilepsy generalized with febrile seizures plus type 7,613863
SCN11A	120.1	99%	99%	Episodic pain syndrome,familial,3,615552 Neuropathy,hereditary sensory and autonomic, type VII,615548
SCN9A	111	100%	99%	Epilepsy generalized with febrile seizures plus type 7,613863 Erythermalgia,primary,133020 Febrile seizures,familial,3B,613863 HSAN2D,autosomal recessive,243000 Insensitivity to pain,congenital,243000 Paroxysmal extreme pain disorder,167400
SEC23B	109	100%	100%	Anemia dyserythropoietic congenital type II,224100
SERPINB7	105.9	100%	100%	Palmoplantar keratoderma, Nagashima type, 615598 ,615598
SERPINB8	102.7	100%	98%	No OMIM disease ID
SERPINH1	129.2	100%	100%	Osteogenesis imperfecta type X,613848 {Preterm premature rupture of the membranes, susceptibility to},610504
SHOC2	116.4	100%	98%	Noonan-like syndrome with loose anagen hair,607721
SKI	63.3	89%	79%	Shprintzen-Goldberg syndrome,182212
SKIV2L	17.3	68%	33%	Trichohepatoenteric syndrome 2,614602
SLC17A9	92.4	95%	93%	Porokeratosis, disseminated superficial actinic, 8,616063
SLC24A4	102.8	98%	95%	Ameliogenesis imperfecta, hypomaturation type, IIA5,615887 [Skin/hair/eye pigmentation 6],210750
SLC24A5	103.6	100%	97%	Albinism, oculocutaneous, type VI,113750 [skin/hair/eye pigmentation 4],113750
SLC26A2	127.1	100%	100%	Achondrogenesis Ib,600972 Atelosteogenesis II,256050 De la Chapelle dysplasia,256050 Diastrophic dysplasia,222600 Diastrophic dysplasia,broad bone-platyspondylic variant,222600 Epiphyseal dysplasia,multiple,4,226900
SLC27A4	75.7	85%	83%	Ichthyosis prematurity syndrome,608649
SLC29A3	147.5	100%	99%	Histiocytosis-lymphadenopathy plus syndrome,602782
SLC2A10	89.4	100%	97%	Arterial tortuosity syndrome,208050
SLC39A13	111.3	100%	99%	Spondylocheirodysplasia Ehlers-Danlos syndrome-like,612350
SLC39A4	69.9	100%	97%	Acrodermatitis enteropathica,201100

SLC45A2	98.9	99%	97%	Oculocutaneous albinism type IV,606574 [skin/hair/eye pigmentation 5],227240
SLC4A4	103.9	100%	100%	Renal tubular acidosis proximal with ocular abnormalities,604278
SLC6A19	81.3	97%	94%	Hartnup disorder,234500 Hyperglycinuria,138500 Iminoglycinuria,digenic,242600
SLC7A7	95	100%	99%	Lysinuric protein intolerance,222700
SLCO2A1	65.8	100%	97%	Hypertrophic osteoarthropathy primary autosomal recessive 2,614441
SLURP1	35.7	96%	84%	Meleda disease,248300
SLX4	128.5	97%	95%	Fanconi anemia complementation group P,613951
SMAD3	72.3	88%	78%	Loeys-Dietz syndrome type 3,613795
SMARCA2	86.4	97%	94%	Nicolaides-Baraitser syndrome,601358
SMARCA4	83.2	98%	92%	Mental retardation autosomal dominant 16,614609 {Rhabdoid tumor predisposition syndrome 2},613325
SMARCAD1	118.9	100%	100%	Adermatoglyphia,136000
SMARCAL1	118.1	99%	97%	Schimke immunoosseous dysplasia,242900
SMARCB1	120.7	100%	100%	Mental retardation autosomal dominant 15,614608 Rhabdoid tumors, somatic,609322 {Schwannomatosis-1,susceptibility to},162091
SMO	100.7	100%	99%	Basal cell carcinoma, somatic
SMOC2	76.5	98%	88%	Dentin dysplasia type I with microdontia and misshapen teeth,125400
SNAI2	79.3	100%	100%	Piebaldism,172800 Waardenburg syndrome, type 2D,608890
SNAP29	118	100%	100%	Cerebral dysgenesis neuropathy, ichtyosis, and palmoplantar keratoderma syndrome,609528
SNRPE	48.6	79%	70%	Hypotrichosis 11,615059
SNX10	96.8	100%	98%	Osteopetrosis autosomal recessive 8,615085
SOS1	113.2	100%	99%	Fibromatosis,gingival,135300 Noonan syndrome 4,610733
SOX10	67.6	100%	100%	PCWH syndrome,609136 Waardenburg syndrome,type 2E,with/without neurological involvement,611584 Waardenburg syndrome,type 4C,613266
SOX18	22.6	76%	44%	Hypotrichosis-lymphedema-telangiectasia syndrome,607823 Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome,137940

SOX2	146.3	100%	100%	Microphthalmia syndromic 3,206900 Optic nerve hypoplasia and abnormalities of the central nervous system,206900
SP7	74.7	100%	100%	Osteogenesis imperfecta type XII,613849
SPINK5	97.3	100%	99%	Atopy,147050 Netherton syndrome,256500
SPINT2	61.2	90%	66%	Diarrhea 3 secretory sodium congenital syndromic,270420
SPRED1	129.3	100%	100%	Legius syndrome,611431
SPRY4	93.8	100%	100%	Hypogonadotropic hypogonadism 17 with or without anosmia,615266
SRD5A3	122.5	100%	100%	Congenital disorder of glycosylation type Iq,612379 Kahrizi syndrome,612713
ST14	85.6	98%	91%	Ichthyosis with hypotrichosis,610765
ST3GAL5	102.4	93%	92%	Ganglioside GM3 synthase deficiency,609056
STAMBP	123.9	100%	100%	Microcephaly-capillary malformation syndrome,614261
STAT3	85	100%	96%	Hyper-IgE recurrent infection syndrome,147060 Autoimmune disease,multisystem,infantile-onset,615952
STAT5B	72.5	83%	75%	Growth hormone insensitivity with immunodeficiency,245590 Leukemia,acute promyelocytic,STAT5B/RARA type
STIM1	81.6	99%	96%	Immune dysfunction with T-cell inactivation due to calcium entry defect 2,612783 Myopathy,tubular aggregate,1,160565 Stormorken syndrome,185070
STK11	73.3	99%	95%	Melanoma malignant, somatic Pancreatic cancer, 260350 Peutz-Jeghers syndrome, 175200 Testicular tumor, somatic, 273300
STS	140.6	100%	99%	Ichthyosis X-linked,308100
SUFU	89.6	98%	90%	Medulloblastoma desmoplastic,155255 Basal cell nevus syndrome,109400 {Meningioma,familial,susceptibility to},607174
SUMF1	72.9	99%	94%	Multiple sulfatase deficiency,272200
TALDO1	95.3	100%	100%	Transaldolase deficiency,606003
TAT	92	100%	98%	Tyrosinemia type II,276600

TBC1D24	109.5	100%	100%	Epileptic encephalopathy early infantile 16,615338 Deafness, autosomal recessive 86,614617 Deafness, autosomal dominant 65,616044 DOOR syndrome,220500 Myoclonic epilepsy,infantile,familial,605021
TBX3	72	98%	92%	Ulnar-mammary syndrome,181450
TCIRG1	73.7	92%	84%	Osteopetrosis autosomal recessive 1,259700
TEK	108	100%	98%	Venous malformations multiple cutaneous and mucosal,600195
TERT	103.7	100%	99%	{Dyskeratosis congenita, autosomal recessive 4},613989 {Dyskeratosis congenita, autosomal dominant 2}, 613979 {Leukemia, acute myeloid},601626 {Melanoma, cutaneous malignant, 9},615134
TFAP2A	72.3	100%	90%	Branchiooculofacial syndrome,113620
TGFB2	123.4	97%	94%	Loeys-Dietz syndrome type 4,614816
TGFBR1	117.5	95%	93%	Loeys-Dietz syndrome type 1A,609192 {Multiple self-healing squamous epithelioma,susceptibility to},132800
TGFBR2	86.2	100%	97%	Colorectal cancer,hereditary,nonpolyposis type 6,614331 Esophageal cancer,somatic,133239 Loeys-Dietz syndrome,type 2,610168
TGM1	101.1	100%	96%	Ichthyosis congenital autosomal recessive 1,242300
TGM5	98	100%	97%	Peeling skin syndrome acral type,609796
TINF2	176.7	100%	100%	Dyskeratosis congenita autosomal dominant 3,613990 Revesz syndrome,268130
TMC6	58.8	99%	94%	Epidermodysplasia verruciformis,226400
TMC8	76.1	99%	94%	Epidermodysplasia verruciformis,226400
TMEM165	85.9	100%	100%	Congenital disorder of glycosylation type IIk,614727
TMEM173	57.9	100%	94%	STING-associated vasculopathy, infantile-onsent (SAVI),615934
TNFRSF11A	100.8	95%	93%	Osteolysis,familial expansile,174810 Osteopetrosis,autosomal recessive 7,612301 {Paget disease of bone 2,early-onset},602080
TNFRSF11B	166.1	100%	100%	Paget disease of bone 5, juvenile-onset,239000
TNFRSF1A	67.7	93%	85%	Periodic fever,familial,142680 {Multiple sclerosis, susceptibility to,5},614810
TNFSF11	131.4	100%	100%	Osteopetrosis,autosomal recessive 2,259710

TNXB	11.3	47%	18%	Ehlers-Danlos syndrome,autosomal dominant,hypermobility type,130020 Vesicoureteral reflux 8,615963
TP63	127.6	100%	100%	ADULT syndrome,103285 Ectrodactyly,ectodermal dysplasia,cleft lip/palate syndrome 3,604292 Hay-Wells syndrome,106260 Limb-mammary syndrome,603543 Orofacial cleft 8,129400 Rapp-Hodgkin syndrome,129400 Split-hand/foot malformation 4,605289
TPCN2	88.1	100%	96%	Skin/hair/eye pigmentation, variation in, 10,612267
TREX1	134.4	100%	100%	Aicardi-Goutieres syndrome 1,dominant and recessive,225750 Chilblain lupus,610448 Vasculopathy,retinal,with cerebral leukodystrophy,192315 {Systemic lupus erythematosus,susceptibility to},152700
TRIM32	103.2	100%	100%	?Bardet-Biedl syndrome 11,615988 Muscular dystrophy,limb-girdle,type 2H,254110
TRIM37	95	100%	98%	Mulibrey nanism,253250
TRPV3	106.6	100%	94%	?Palmoplantar keratoderma,nonepidermolytic,focal 2,616400 Olmsted syndrome,614594
TSC1	89.9	99%	97%	Focal cortical dysplasia,Taylor balloon cell type,607341 Lymphangioleiomyomatosis,606690 Tuberous sclerosis-1,191100
TSC2	81.8	98%	95%	Lymphangioleiomyomatosis,somatic,606690 Tuberous sclerosis-2,613254
TTC37	111.4	100%	100%	Trichohepatoenteric syndrome 1,222470
TTI2	100.5	99%	97%	Mental retardation, autosomal recessive 39,615541
TWIST2	79.7	100%	99%	Ablepharon-macrostomia syndrome,200110 Barber-Say syndrome,209885 Focal facial dermal dysplasia 3,Setleis type,227260
TYR	138	74%	74%	Albinism,oculocutaneous,type IA,203100 Albinism,oculocutaneous,type IB,606952 Waardenburg syndrome/albinism,digenic,103470 [Skin/hair/eye pigmentation 3],601800
TYRP1	106.5	100%	99%	Albinism oculocutaneous type III,203290 [Skin/hair/eye pigmentation,variation in,11(Melanesian blond hair),612271

UBE2A	101.2	100%	100%	Mental retardation,X-linked syndromic,Nascimento-type,300860
UBR1	101	100%	100%	Johanson-Blizzard syndrome,243800
UROD	84.4	99%	91%	Porphyria,cutanea tarda,176100 Porphyria,hepatoerythropoietic,176100
UROS	81.4	95%	83%	Porphyria,congenital erythropoietic,263700
USB1	52.1	91%	83%	Poikiloderma with neutropenia,604173
UVSSA	65.5	100%	95%	UV-sensitive syndrome 3,614640
VDR	77.4	100%	99%	Rickets,vitamin D-resistant,type IIA,277440 ?Osteoporosis,involutional,166710
VEGFC	87.2	99%	97%	Lymphedema, hereditary, ID,615907
VHL	114.9	100%	100%	Erythrocytosis,familial,2,263400 Hemangioblastoma,cerebellar,somatic Pheochromocytoma,171300 Renal cell carcinoma,somatic,144700 von Hippel-Lindau syndrome,193300
VPS13B	104.4	99%	98%	Cohen syndrome,216550
WAS	60.1	100%	90%	Neutropenia,severe congenital,X-linked,300299 Thrombocytopenia,X-linked,313900 Wiskott-Aldrich syndrome,301000
WDR19	115.8	100%	99%	?Cranioectodermal dysplasia 4,614378 ?Short-rib thoracic dysplasia 5 with/without polydactyly,614376 Nephronophthisis 13,614377 Senior-Loken syndrome 8,616307
WDR35	108.7	100%	98%	Cranioectodermal dysplasia 2,613610 Short-rib thoracic dysplasia 7 with/without polydactyly,614091
WDR72	114.7	99%	99%	Amelogenesis imperfecta hypomaturation type IIA3,613211
WIPF1	98.2	97%	94%	Wiskott-Aldrich syndrome 2,614493
WNT10A	68.9	94%	88%	Odontoonychodermal dysplasia,257980 Schopf-Schulz-Passarge syndrome,224750 Tooth agenesis,selective,4,150400
WNT10B	90.7	100%	93%	Split-hand/foot malformation 6,225300
WNT5A	111.3	100%	98%	Robinow syndrome autosomal dominant,180700
WNT7A	129.9	100%	100%	Fuhrmann syndrome,228930 Ulna and fibula,absence of,with severe limb deficiency,276820

WRAP53	142.8	100%	98%	Dyskeratosis congenita autosomal recessive 3,613988
WRN	126.7	100%	98%	Werner syndrome,277700
XPA	80.9	100%	93%	Xeroderma pigmentosum group A,278700
XPC	114.6	99%	97%	Xeroderma pigmentosum group C,278720
XYLT1	104.5	95%	89%	Desbuquois dysplasia 2,615777 {Pseudoxanthoma elasticum,modifier of severity of},264800
XYLT2	80.5	94%	91%	Spondyloocular syndrome,605822 {Pseudoxanthoma elasticum,modifier of severity of},264800
ZBTB20	134	100%	100%	Primrose syndrome,259050
ZMPSTE24	137.9	100%	100%	Mandibuloacral dysplasia with type B lipodystrophy,608612 Restrictive dermopathy,lethal,275210
ZNF469	92	100%	99%	Brittle cornea syndrome 1,229200
ZNF592	107.3	93%	91%	Spinocerebellar ataxia,autosomal recessive 5,606937
ZNF750	120.6	100%	100%	Seborrhea-like dermatitis with psoriasiform elements,610227

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.
Median Coverage describes the average number of reads seen across 50 exomes
% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x
% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x
OMIM release used for OMIM disease identifiers and descriptions : November 15th, 2015
This list is accurate for all panel versions starting with DG 2.4. (where x is a random number signifying a minor analysis patch without consequences for the panel composition or coverage information)

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors